



Czech dysplasia

Czech dysplasia is an inherited condition that affects joint function and bone development. People with this condition have joint pain (osteoarthritis) that begins in adolescence or early adulthood. The joint pain mainly affects the hips, knees, shoulders, and spine and may impair mobility. People with Czech dysplasia often have shortened bones in their third and fourth toes, which make their first two toes appear unusually long. Affected individuals may have flattened bones of the spine (platyspondyly) or an abnormal spinal curvature, such as a rounded upper back that also curves to the side (kyphoscoliosis). Some people with Czech dysplasia have progressive hearing loss.

Frequency

The prevalence of Czech dysplasia is unknown; at least 11 families have been affected. Most of these families reside in the Czech Republic.

Genetic Changes

Czech dysplasia is caused by a particular mutation in the *COL2A1* gene. The *COL2A1* gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in the clear gel that fills the eyeball (the vitreous) and in cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Type II collagen is essential for the normal development of bones and other connective tissues that form the body's supportive framework. Mutations in the *COL2A1* gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from developing properly.

Inheritance Pattern

Czech dysplasia is inherited in an autosomal dominant pattern, which means one copy of the altered *COL2A1* gene in each cell is sufficient to cause the disorder.

All known individuals with Czech dysplasia inherited the mutation from a parent with the condition.

Other Names for This Condition

- Czech dysplasia, metatarsal type
- progressive pseudorheumatoid dysplasia with hypoplastic toes
- spondyloarthropathy with short third and fourth toes

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Czech dysplasia metatarsal type
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1836683/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Connective Tissue Disorders
<https://medlineplus.gov/connectivetissuedisorders.html>
- Health Topic: Osteoarthritis
<https://medlineplus.gov/osteoarthritis.html>

Genetic and Rare Diseases Information Center

- Czech dysplasia metatarsal type
<https://rarediseases.info.nih.gov/diseases/10220/czech-dysplasia-metatarsal-type>

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Arthritis
https://www.niams.nih.gov/Health_Info/Osteoarthritis/osteoarthritis_ff.asp
- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Connective Tissue
https://www.niams.nih.gov/Health_Info/Connective_Tissue/

Educational Resources

- Disease InfoSearch: Czech Dysplasia Metatarsal Type
<http://www.diseaseinfosearch.org/Czech+Dysplasia+Metatarsal+Type/2089>
- MalaCards: czech dysplasia
http://www.malacards.org/card/czech_dysplasia
- Orphanet: Czech dysplasia, metatarsal type
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=137678

Patient Support and Advocacy Resources

- International Skeletal Dysplasia Registry, UCLA
<http://ortho.ucla.edu/isdr>
- Resource list from the University of Kansas Medical Center: Hard of Hearing
<http://www.kumc.edu/gec/support/hearing.html>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28czech+dysplasia%5BTIAB%5D%29+OR+%28progressive+pseudorheumatoid+dysplasia+with+hypoplastic+toes%29+OR+%28spondyloarthropathy+with+short+third+and+fourth+toes%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- CZECH DYSPLASIA
<http://omim.org/entry/609162>

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